

## Scoliosis in Neurofibromatosis, Type 1

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About one third of all patients with NF1 will have issues affecting their spine sometime in their lifetime. Among the skeletal issues in NF1, scoliosis is by far the most serious and may have a significant lifelong impact. Scoliosis in NF1 may occur in 3 different forms: adolescent idiopathic scoliosis, scoliosis secondary to a paraspinous tumor, and dystrophic scoliosis. In this brief article, we will discuss NF1-associated scoliosis and its appropriate management.

### Scoliosis defined

Scoliosis is defined as an abnormal curvature of the spine. The spine is made of three parts: the cervical spine, the thoracic spine and the lumbar spine. The sacrum and coccyx are below the spine (what we know as the “tailbone”). The normal spine does exhibit “normal” curves. When the spine curves toward the front, it is called **lordosis**. When it curves toward the back, it is called **kyphosis**. These front and back curves are expected, but side-to-side curves are not normal. Mild curves are well tolerated, but a sideways curve of more than 10 degrees may become problematic. These curves may become progressive, leading to pain, limited mobility and, in severe cases, breathing or heart problems.



Source: spineuniverse.com

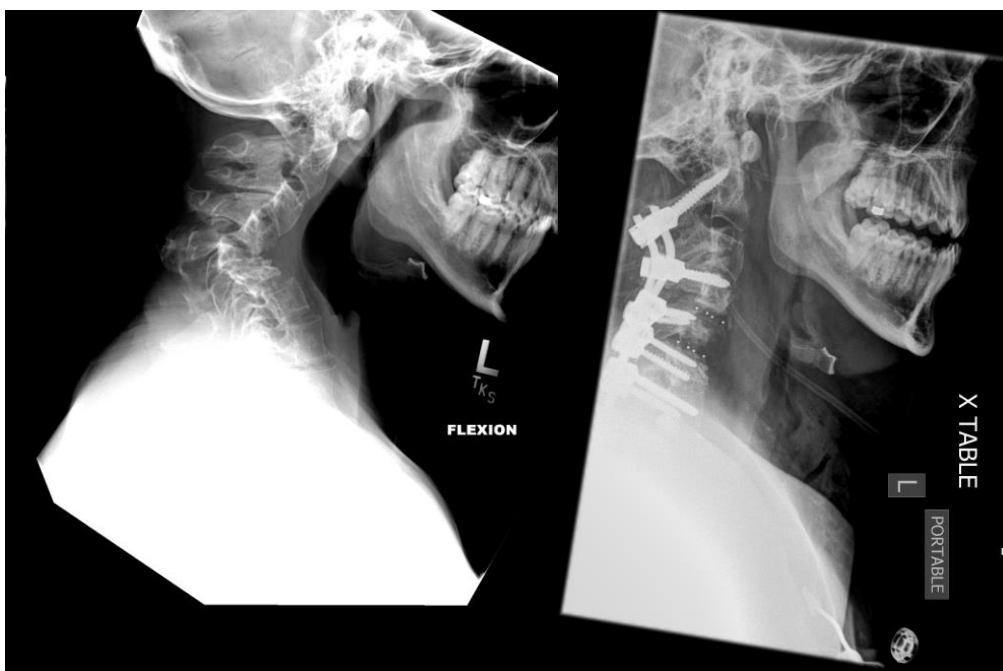
### Scoliosis in NF1

We know that people with NF1 have an increased incidence of scoliosis. It is always important for your caregiver to screen for scoliosis at all ages. In patients with and without NF1, the most common form of scoliosis appears during adolescence. This is a period of significant growth during which any imbalance in the spine can become exaggerated. This can happen in NF1 also, but there are individuals with NF1 who can develop scoliosis at any age. This is called “dystrophic scoliosis”, a severe form of scoliosis that

results from abnormal bone development in some patients with NF1. This form of scoliosis can come on quickly and progress to a severe form. Dystrophic scoliosis may exhibit the following features:

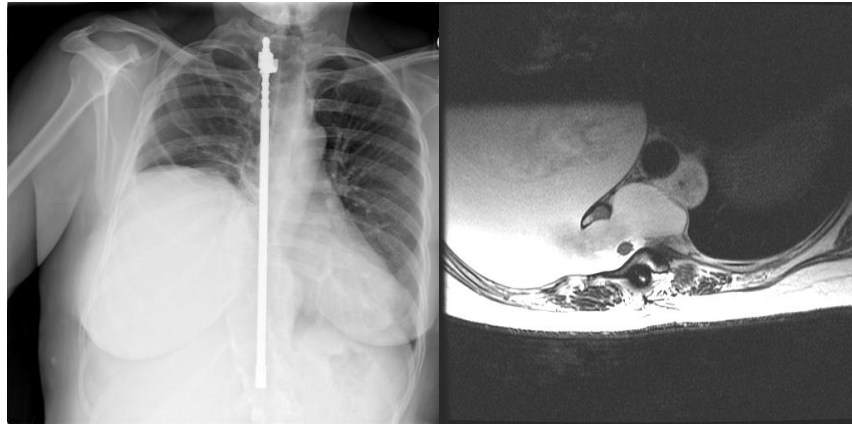
1. Vertebral wedging. Instead of being essentially square or rectangular in shape, the main bones of the spine become almost triangular causing the spine to grow out of alignment.
2. Atypical curve/location. Adolescent scoliosis usually involves the thoracolumbar spine. Dystrophic scoliosis may involve the cervical, thoracic or lumbar spine in isolation. The curve can also be more severe and angulated.
3. Vertebral rotation is another way the dystrophic spine grows out of alignment in any direction.
4. Rib penciling – remodeling of the head of the rib associated with scoliotic curves.

The X-rays below show a patient before and after surgery with dystrophic scoliosis of the cervical spine (atypical curve/location) with severe vertebral wedging.

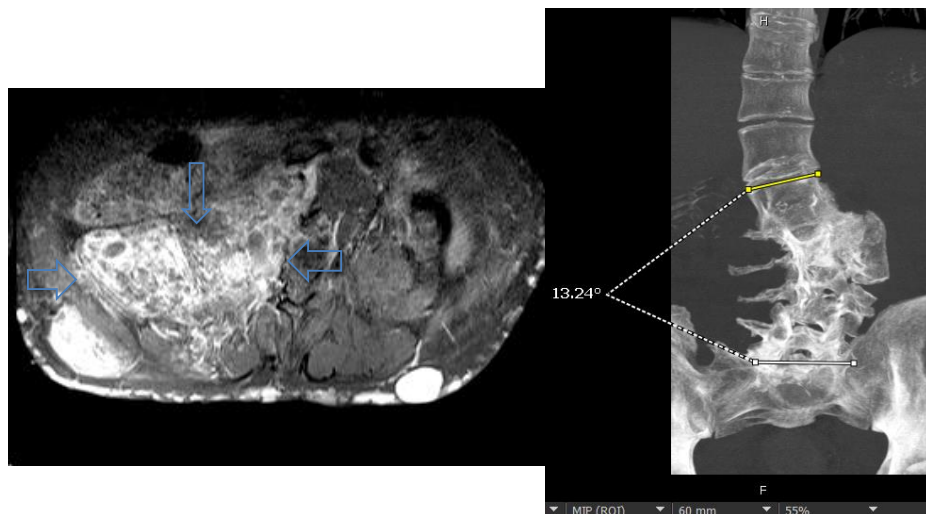


Someone with a great deal of training and experience with this patient group should always perform repair of spinal deformities in patients with NF1. Any scoliosis repair should also bear in mind that bones in patients with NF1, especially those with dystrophic scoliosis, behave differently than bones in patients in the general population. There is active research going on to find out ways to improve bone formation in NF1 and to improve the outcome of repair for conditions like dystrophic scoliosis. These repairs can break down years later, so part of regular medical care for patients who had spine surgery includes regular check ups by a NF specialist or spine surgeon.

The pictures below are from a woman who had spine surgery for NF1 many years earlier, but the bones at the site of her repair had since worn away and allowed fluid from inside the spine to go out and fill her chest. She had not had regular follow-up, but showed up in the NF clinic with shortness of breath!



If a new spinal curve appears rather suddenly, one should always keep in mind that this may be the result of a new tumor. In NF1, this is especially important since neurofibromas next to the spine can also cause the spine to grow abnormally. The picture on the left below shows a large neurofibroma seen on an MRI. The picture to the right shows the resulting spinal deformity.



Finally, adults, adolescents and children contemplating spinal surgery should know it hurts! A pain management plan should be in place before surgery is performed so that pain is well controlled and inappropriate use of narcotics is avoided.

Active research continues regarding bone development and the contributing factors that cause scoliosis in NF1. We have looked at pieces of DNA that may predict who is at greatest risk for scoliosis. Someday, there may be a way to accurately predict scoliosis risk from a cheek swab! It also appears that the dysregulation of RAS in NF1 is a major contributor to bone loss. The new class of RAS pathway inhibitors may help to return bone metabolism to normal. A group of drugs called bisphosphonates, which are very helpful for people with osteoporosis, may also be helpful for people with NF1. We know that vitamin D is very helpful for bone health, so spending a moderate amount of time in the sun and taking supplemental vitamin D as part of a healthy diet (especially for those who live in northern states or countries) should be part of the routine for all patients with NF1. Finally, we know that regular exercise contributes to bone health; so walk, swim, run, be active!